

ABSTRACT OF THE DISCLOSURE

This invention relates generally to the gene, and mutations, that are responsible for the disease hemochromatosis (HH). In particular, the present invention provides for the presence of one or more mutations on the ferroportin 1 (SLC11A3) gene which results in aberrant SLC11A3 mediated iron transport. The invention also relates to methods for diagnostic tools, drugs and therapies developed for the treatment of patients with HH or anemia.

DEPT. OF STATE 2610